CHARGE syndrome after 35 years
Developmental & Educational Aspects

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CDBS SOCAL CHARGE Day
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Resources & literature
- CHARGE Syndrome Foundation (Professional Packet, AJMG, ASHA video, Parent Manual, CHARGE Accounts, webinars, links)
- Perkins School for the Blind CHARGE webcasts
- CHARGE Lab at Central Michigan University
- Bulldog CHARGE Lab in Mississippi
- DB-LINK
- Texas School for the Blind & Visually Impaired
- California Deafblind Services
- Sense UK CHARGE Information Packet
- Books from the USA, from the CHARGE Association of Australasia, and from Germany
- Face Book

American Journal of Medical Genetics
Volume 175C Number 4 December 2017

- New insights and advances
- Genetic counseling
- Genotype analysis of 119 French patients
- Behaviour
- Inner ear malformations
- Cranial imaging
- Cerebellar foliation anomalies
- Neural crest defects
- The heart
- Gastrointestinal and feeding difficulties
- Endocrine phenotypes
- Immunodeficiency

CHARGE Lab research topics
- The effects of a fun chi video on sleep and emotional self-regulation in children with charge syndrome
- Interventions for sleep problems in CHARGE syndrome
- The development of social play in CHARGE syndrome
- Profiles of ability in CHARGE syndrome
- Anxiety in CHARGE syndrome
- Restorative environments for CHARGE syndrome
- Headaches in CHARGE syndrome
- Checklist of educational issues in CHARGE syndrome
- Post-traumatic growth in parents of children with CHARGE syndrome
- The effects of psychotropic medication on children with CHARGE syndrome

Bulldog CHARGE Lab research topics
- CHARGE and IEPs
- CHARGE and family demographics
- CHARGE and issues of sexuality
- Differences between CHARGE and autism

The changing nature of the population of children with CHARGE syndrome
1981 - 6 anomalies
• C - Coloboma
• H - Heart Defects
• A - Atresia of the Choanae
• R - Retarded Growth/Development
• G - Genito-urinary Defects
• E - Ear anomalies

2005 - 38 anomalies???
• Mutation of gene CHD 7
• Facial Palsy/facial clefting
• Cranial Nerve anomalies
• Semi-circular canal anomalies
• Dental anomalies
• Sense of smell anomalies
• Seizure disorder
• Immune deficiency
• Sleep apnea

Dental Issues
• Delayed (or early) eruption
• Teeth grinding
• Poor quality dental enamel
• Pouching of food
• Medications
• Reflux and vomiting
• Facial clefting
• Facial palsy
• Misaligned teeth

Cranial Nerve Anomalies
• Nerve 1 - Smell (42%)
• Nerve 2 - Vision (80%)
• Nerve 7 - The Face (43%)
• Nerve 8 - Hearing & Balance (80%)
• Nerves 9 and 10 - Swallowing (50%)

(From CHARGE Foundation Parent Manual - 1999)

What usually gets overlooked?
• Dental issues
• Hypocalcaemia
• Hypoglycaemia
• Seizures
• Retinal detachments
• Constipation
• Obstacles to walking
• Obstacles to speaking/signing
• Scoliosis
• Immune system deficiency
• Self-regulation

CHARGE syndrome involves many more senses than just vision & hearing, and it is not enough only to consider the tactile sense as a compensatory channel.
Deafblindness/multi sensory impairment

“Most people with CHARGE that I have met satisfy the criteria for being considered as having deaf-blindness, even if they have some useful vision and hearing. This is a disability that is defined in functional, not clinical, terms and for each individual with CHARGE it is mostly about difficulties in accessing information not just from the world around them but even from their own bodies.”

David Brown (2011) Deaf-Blindness, Self-Regulation, and Availability for Learning: Some Thoughts on Educating Children with CHARGE Syndrome reSources Volume 16 Number 3

“Brown (2011) argued that a deafblind pedagogy is likely to provide the ‘best fit’ when educating learners with CHARGE, although he also suggests that there is the likelihood or need to consider other issues that might be more specific to this group of learners.” p3

“The view that deafblind pedagogy might provide the ‘best fit’ for learners with CHARGE was also explored. The findings of this investigation suggest that the deafblind educational philosophy and approaches to teaching are likely to be an effective pathway to follow for educators of learners with CHARGE. In addition, however, it was found that some aspects of established deafblind practice may need to be applied in an alternative way or for a different purpose, and that a supplementary approach is also likely to be required.” p14-15

Gail Deuce “The Education of Learners with CHARGE Syndrome” British Journal of Special Education 2017

“Children with CHARGE syndrome are truly “multi-sensory impaired”, having difficulties not only with vision and hearing but also with the senses that perceive balance, touch, temperature, pain, pressure, and smell, as well as problems with breathing and swallowing, eating and drinking, digestion, and temperature control.”


Personal conclusion after more than 30 years of scientific and practical work

“The multi-sensory impaired person is a unique human being with a unique line of development, who is more dependent on the professional’ s willingness to accept this and act accordingly than any other group of disabled persons.”


“Even though it is not totally possible, we must try to step into the experience of our students and how they perceive the world. By doing so, we will find the best ways to communicate together and to create meaningful learning opportunities that will open up the world to them.”

Marianne Riggio Deafblind International Review #56 January 2016
“A good and effective educational program, while being very positive and having high expectations, should always take account of the fact that everything that a child with CHARGE syndrome does is likely to take more thought, and attention, and concentration, and energy, and time for them than it does for us.”

David Brown (2011) Deaf-Blindness, Self-Regulation, and Availability for Learning: Some Thoughts on Educating Children with CHARGE Syndrome reSources Volume 16 Number 3

CHARGE - the most ‘multi sensory impaired’ of all syndromes
Problems with the perception of:

• Vision
• Hearing
• Touch
• Proprioception
• Temperature
• Pain
• Vestibular
• Smell
• Taste

Most people focus on the child’s disabilities, but close attention to their abilities, and to the things that they do, can reveal more about the difficulties they face and the strategies they use to function effectively.

Everything that children with CHARGE do has meaning, and the first obligation on the teacher is to ascertain that meaning (or at least to come up with a really good guess).

“The Forgotten Senses”

PROPRIOCEPTION
The receptors are in the muscles and joints throughout the body

Tells us about the position of our body and all of our limbs, and if anything is moving

VESTIBULAR
The receptors are in the Inner Ears

Tells us about head position & the pull of gravity, detects motion, and it has very close links with the eyes and vision

PROPRIOCEPTION

When the proprioceptive sense is not working properly one common outcome may be:

Seeking strong pressure, stretching, or twisting inputs, eg. squeezing into tight spaces, crossing or twisting limbs around each other, twisting a foot or a leg around the leg of a chair, binding parts of the body with cloth or string or rubber bands, pulling downwards on the teeth and lower jaw, grinding the teeth, tapping the teeth, hand clapping or flapping, leg swinging or kicking, hanging from a bar, jumping up and down, banging the head, hammering objects, standing on the head.
“Of all the many sensory impairments associated with CHARGE syndrome, absence of, or significant damage to, the vestibular sense is perhaps the most far-reaching in its implications, the least understood, and the most overlooked.”


“In the future it is likely that we will discover close links between significant vestibular impairment and many of the currently ‘unexplained’ CHARGE behavioural features such has difficulty with the self-regulation of arousal levels, sudden and apparently unpredictable mood changes, poor memory, and executive dysfunction.”


“A substantial portion of the body of knowledge about the consequences of vestibular dysfunction in CHARGE has been obtained by perceptive observation and not with objective tests. However, objective tests often provide information about the dysfunction of specific structures that may be amenable to treatment.”

Thelin, Curtis, Fussner Maddox, Travis “Balance and Mobility” in Hartshorne, Hefner, Davenport, Thelin (2011) “CHARGE Syndrome” Chapter 6, pp 60

Jean Ayres (1979)
Sensory Integration and the Child

“The vestibular system is the unifying system. All other types of sensation are processed in reference to this basic vestibular information. The activity in the vestibular system provides a framework for the other aspects of our experiences.”

Dr. George Williams “Balance in CHARGE”
CHARGE Syndrome Foundation Manual

Vestibular function has a role in...
- Detecting motion
- Detecting & responding to gravity
- Providing stability during body movement
- Locating body parts & developing body schema
- Influencing muscle tone and posture
Facilitating the crossing of the midline
• Motor control, coordination & sequencing
• Assisting with auditory & visual perception
• Modulating arousal & alertness for attention and calming

Early effects of poor (or missing) vestibular function in children with CHARGE Syndrome
(Brown, American Journal of Medical Genetics, March 2005)
• Low muscle tone (“floppy muscles”)
• Poor head control & poor ability to resist gravity
• Strong postural insecurity when held upright
• Marked preference for being flat on the back
• Delayed mobility & unusual movement patterns (eg. back scooting, side-winding, five-point crawl)

Early effects of poor (or missing) vestibular function in children with CHARGE Syndrome (Brown, cont.)
• “W” sitting for broader, more secure base
• Better visual, auditory and fine motor skills in supine than in the upright position
• Poor bilateral coordination, hand and eye dominance is either total or missing
• Fatigue after trying to resist gravity for periods of time
• Difficulties with self-regulation

“For children with CHARGE syndrome gravity sucks”
Maryann Girardi, Physical Therapist at Perkins School for the Blind

Early mobility
• Rolling
• Side-winding
• Back scooting
• Bottom shuffling
• 5-point crawling
• Regular crawling

The Equilibrium Triad
- Touch/Proprioception
- Vision
- Vestibular
Walking
• Walkers
• Walking poles
• Rolling gait
• Flat feet/ bent knees
• Foot slapping
• Tip-toe walking
• Feet roll inwards/ knees knock together
• Arms extended to the sides or in front
• Fingers crossed/hands clenched
• Eyes fixed on a visual target ahead (holding on with the eyes)

“Children often adopt specific postures (e.g., horizontal with both legs bent and one ankle up crossing the other knee, or legs tightly crossed, or fingers crossed or bunched together, or hands fist ed, or arms folded). These postures provide essential extra tactile and pressure information to the brain about where the child’s limbs are in space, and also confirms for them that they are securely “fixed” and not moving or floating around.”

These postural behaviours have always been present but largely unseen. When they are noticed there is often amusement because the postures seem weird, or the child is corrected with no attempt to understand or recognise or honour what the posture means and what function it serves.

The brain is connected to the body through the senses

I believe that most children with CHARGE syndrome are not in touch with/ do not feel their bodies very well

Before and after walking has been attained we commonly see postures involving…..
• the head
• the legs
• bending/stretching
• hanging
• the hands
• planking
• propping
• squeezing
• climbing

Vision and the vestibular sense
You fix the body......
to fix the head......
to fix the eyes......
so you can use your vision in the best, most reliable, and most comfortable way possible.

“After air to breathe, postural security is our next most urgent priority.”

Jean Ayres

Functions of these postures
• To locate the body (especially the head)
• To confirm postural security
• To stabilise the body, to stabilise the head, to stabilise the eyes for visual tasks
• To ease the discomfort of chronic constipation
• To open up the airway
• To self regulate

What helps? (1)
• Advice from PT and OT
• Activities which improve strength, muscle tone, and controlled movement, and reinforce the body/brain connection (eg. Tai Chi, yoga, wrestling, climbing, dancing, APE, horse riding)
• Deep pressure inputs (eg. jumping, massage, swimming)
• Binding (eg. spandex pressure vest)
• Good physical support & appropriate postures for efficient functioning

“Spandex is the best friend of children with CHARGE syndrome”

Maryann Girardi, Physical Therapist at Perkins School for the Blind
What helps? (2)
• Variety in postures and movement
• Rest periods for re-organization
• Controlled environments
• Self-taught and taught strategies
• Strategies that are motivational
• Appropriate vocabulary (for body parts, for physical feelings, for emotional states, for desired activities)

Yes, I believe that posture should be included as a “self-stimulation” and/or a “self-regulation” behavior (especially for people with CHARGE syndrome)

BREAKOUT SESSIONS in Orlando, August 2017
Lauren Liebermann et al “Promoting Recreation and Physical Activity in the Transition Process”
Sarah Bis & Sarah Maust “Managing the Sensory Environment”
Elizabeth Foster “Quality APE & Recreation for Individuals with CHARGE Syndrome”
Elizabeth Foster “Variables that Impact Walking in Individuals with CHARGE Syndrome”
Michael Braga “Purposeful play: how wrestling and playing can benefit children with CHARGE”
Kate Beals “Connect the dots”

POSTER SESSIONS in Orlando 2017
The Sensory Base of Unusual Behaviors in Children with CHARGE
Trends in and Barriers to Recreation Participation in CHARGE Syndrome
Impact of Variables on the Attainment of Walking in Children with CHARGE Syndrome
Effective Positioning Interventions to Facilitate Fine Motor Skills in School-Aged Children with CHARGE Syndrome and/or deafblindness
Teaching Gross Motor Skills and Balance to Children with CHARGE Syndrome

*Communication with one’s own body
*Communication with one’s immediate environment
*Communication with the wider world

Communication Options
- Objects & calendars
- Sign language
- Speech with hearing aids
- Speech with a cochlear implant
- Visual programs
- Signed English
- Reading and writing
The way to success?.....
.....the child’s preferred mode(s) of communication

“Many children, if they have a range of communication options available to them, both receptively and expressively, clearly show their skill at choosing what suits them best from moment to moment.”

David Brown (2011) Deaf-Blindness, Self-Regulation, and Availability for Learning: Some Thoughts on Educating Children with CHARGE Syndrome reSources Volume 16 Number 3

Health issues
Complex health issues
Availability of medical/para-medical services
Balancing health and educational needs
Making health care educational

CHARGE management issues
• Constipation - Autonomic nervous system?
• Toilet training delays– nerve abnormalities?
• Immune system deficiency
• Sleep disturbances - abnormal circadian cycle
• Cyclic vomiting/abdominal migraines
• Sensory integration issues
• Behaviors, especially self-regulation issues

What is CHARGE Behavior??
• Impulsivity
• High anxiety
• Obsessions
• Self stimulation
• Poor self-regulation
• Executive function disorder
• Attention Deficit Hyperactivity Disorder
• Pervasive Developmental Disorder
• Obsessive Compulsive Disorder
• “Autistic-like” behaviors
• Deafblindness
• Multi-sensory problems……(and so on!)
• Laziness, stubbornness, aggression

[Self-regulation]… “is defined as the capacity to manage one’s thoughts, feelings and actions in adaptive and flexible ways across a range of contexts”
Jude Nicholas, CHARGE Accounts, Summer 2007
behavioral difficulties, some of which may be described as autistic-like, and obsessive-compulsive, with attention difficulties... There are potentially multiple sources for these difficulties... Multi-sensory impairments, communication frustrations, and physical pain and discomfort all have been implicated. However, some children with fairly good sensory abilities, adequate communication, and little apparent discomfort may still have challenging behavior. Cognitive impairment has been implicated in some but not all cases. It seems likely that some neuropsychological problems exist. Recent research supports the presence of executive dysfunction, or problems with shifting, initiating, inhibiting, or sustaining actions... Another area being explored is the presence of a regulatory disorder making it difficult for the child to regulate complex processes such as their sleep-wake cycle, hunger-satiety cycle, their ability to console themselves, to manage their emotions, and to plan their motor activities.

Tim Hartshorne, CHARGE Foundation Professional Packet

Sensory Integration Issues
- Extreme sensitivity (or under reaction) to touch, movement, sights, or sounds
- Distractibility
- Social and/or emotional problems
- Activity level that is unusually high or unusually low
- Physical clumsiness or apparent carelessness
- Impulsivity, or lack of self-control
- Difficulty making transitions from one situation to another
- Inability to unwind or calm one’s self
- Delays in speech, language, or motor skills
- Delays in academic achievement

The Future??
- Medical research & advances - genetics, immunology, digestive system, osteoporosis, vestibular issues, sleep disturbance, low muscle tone
- Improved identification and access to services
- Age-related developments, both medical and behavioural
- Investigation of behavioural issues, especially self-regulation & executive dysfunction
- Improved awareness of the correct assessment procedures and educational strategies

David Brown on CHARGE - 1997
“I know of no identified sub-group within the population of people with multi sensory impairment who have so many medical problems, of such complexity and severity, and with so many hidden or delayed difficulties, and yet no sub-group has shown such a consistent ability to rise triumphantly above these problems”

“Exploring executive functioning”-Amanda Kirby
- Activation – organizing & prioritizing, initiating, getting started
- Focus – sustaining & shifting, completing
- Effort – regulating alertness
- Emotion – managing frustrations, modulating emotions, keeping perspective
- Memory – remembering, accessing recall, recognizing & remembering a sequence
- Action – monitoring & regulating self-action without impulsivity, or poor context or poor pacing

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